KCNJ5 gene

potassium voltage-gated channel subfamily J member 5

Normal Function

The *KCNJ5* gene provides instructions for making a protein that functions as a potassium channel, which means that it transports positively charged atoms (ions) of potassium into and out of cells. Potassium channels produced from the *KCNJ5* gene are thought to help regulate production of the hormone aldosterone. In the adrenal glands, which are small hormone-producing glands located on top of each kidney, the channels control the flow of ions into the cell. The flow of ions creates an electrical charge across the cell membrane, which affects the triggering of certain biochemical processes (pathways) that regulate aldosterone production. Aldosterone helps control blood pressure by maintaining proper salt and fluid levels in the body.

Health Conditions Related to Genetic Changes

familial hyperaldosteronism

At least four *KCNJ5* gene mutations have been identified in people with familial hyperaldosteronism type III. Familial hyperaldosteronism is a disorder that causes high blood pressure (hypertension). Mutations in the *KCNJ5* gene are thought to result in the production of potassium channels that are less selective, allowing other ions (predominantly sodium) to pass as well. The flow of sodium ions into adrenal gland cells affects the electrical charge across the cell membrane, activating another type of channel that allows calcium ions to enter. The influx of calcium ions overactivates a process called the calcium/calmodulin pathway that increases aldosterone production, resulting in excess aldosterone and the hypertension associated with familial hyperaldosteronism type III.

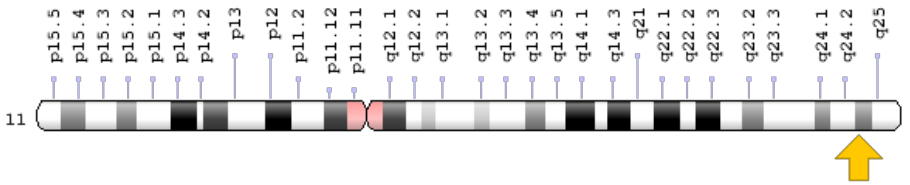
other disorders

Some gene mutations are acquired during a person's lifetime and are present only in certain cells. These changes, which are called somatic mutations, are not inherited. Somatic mutations in the *KCNJ5* gene have been identified in approximately 40 percent of nonhereditary (sporadic) tumors of the adrenal glands called aldosterone-producing adenomas. These noncancerous (benign) tumors cause hypertension that gets worse over time. As in familial hyperaldosteronism (described above), *KCNJ5* gene mutations in cells of the adrenal gland result in increased aldosterone production, leading to hypertension.

Chromosomal Location

Cytogenetic Location: 11q24.3, which is the long (q) arm of chromosome 11 at position 24.3

Molecular Location: base pairs 128,891,418 to 128,921,163 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cardiac ATP-sensitive potassium channel
- CIR
- G protein-activated inward rectifier potassium channel 4
- GIRK4
- heart KATP channel
- inward rectifier K+ channel KIR3.4
- IRK-4
- KATP1
- KIR3.4
- LQT13
- potassium channel, inwardly rectifying subfamily J, member 5
- potassium inwardly-rectifying channel, subfamily J, member 5

Additional Information & Resources

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28KCNJ5%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 5
  http://omim.org/entry/600734

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_KCNJ5.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=KCNJ5%5Bgene%5D
- HGNC Gene Family: Potassium voltage-gated channel subfamily J
  http://www.genenames.org/cgi-bin/genefamilies/set/276
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6266
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3762
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